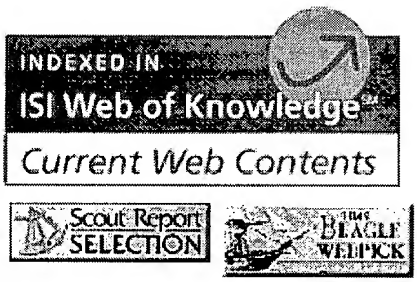
Search results

MITOMAP

A human mitochondrial genome database

A compendium of polymorphisms and mutations of the human mitochondrial DNA



Search MITOMAP for information on: [ ] Perform search Clear

Gene, disease, enzyme names may be abbreviated, truncated, etc. Examples of search words: ND1, ND4, NARP, LHON, 11778, 3243, Leu, Lys, etc.

Mitomap Quick Reference

The Human Mitochondrial Sequence (updated)

Amino Acid Translation Tables

Mitochondrial References (A-Z) (>1 MB)
or view only authors A-L or authors M-Z

Mitochondrial DNA Function Locations (Gene Loci)

Mitochondrial DNA Polypeptide Assignments

Polymorphic mtDNA Restriction Sites (High Resolution Screening)

Common Continent-Specific mtDNA Variants

Mitochondrial Human Genome Report

Illustrations
- -Mitochondrial DNA Map
- -Eleven pathological mutations in $tRNA^{Leu(UUR)}$
- -Mitochondrial energetics
- -Diabetes metabolism & the mitochondria
- -World migrations
- -mtDNA Tree

Other databases:
HmtDB Population & Biomedical Database
mtDB Database
FBI Forensic mtDNA Database
Human Mitochondrial Protein Database
Mammalian Mitochondrial tRNA Genes
DNA Polymerase Gamma Mutation Database
eOPA1: OPA1 Mutation Database

Other Useful Links on the Web

The following web pages are refreshed regularly from the database:

- MtDNA Polymorphisms (includes mini insertions & deletions)

  Control Region Polymorphisms (16024-576)
  Coding & RNA Polymorphisms (577-16023, MTTF-MTTP)
  Somatic Mutations

Collection of Unpublished Polymorphisms/Mutations

mtDNA Tree Tree Bibliography

- MtDNA Mutations with Reports of Disease-Associations

Organized by mtDNA location:	Organized by phenotype:
rRNA/tRNA Mutations	rRNA/tRNA Mutations
Coding & Control Region Mutations	Coding & Control Region Mutations
	LHON Mutations

- Major Rearrangements

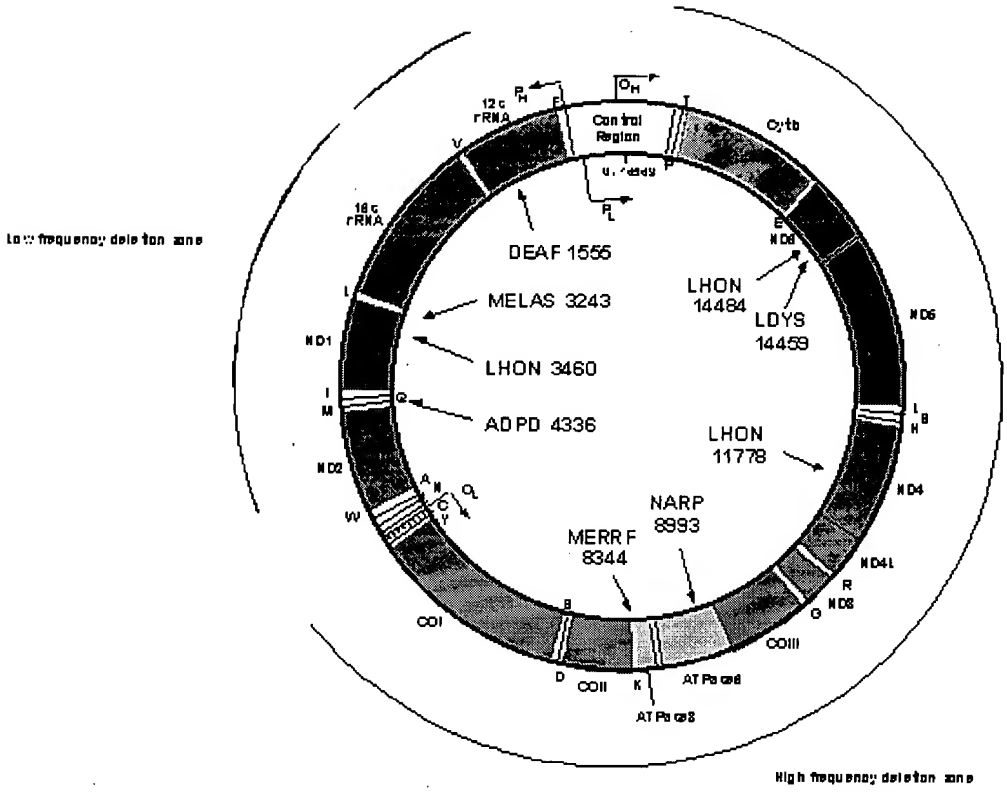
MtDNA Deletions
Multiple mtDNA Deletions Within Individuals
MtDNA Inversions
MtDNA Simple Insertions
MtDNA Complex Rearrangements

- Nuclear Genes Involved in Mitochondrial Disease

Structural Nuclear Genes in Mitochondrial Disease
Non-Structural Nuclear Genes in Mitochondrial Disease

- Mitochondrial Pseudogenes

- Submittal form for unpublished mtDNA polymorphisms
- Submittal of articles & published data: If you would like to submit published articles to be included in mitomap, please send the citation & a pdf file to mitocite@uci.edu .
- Complete Mitochondrial Genome Sequences
- Mitochondria Interest Group Video Casts
- mtDNA in the news: NY Times on the Web, May 2, 2000 "The Human Family Tree: 10 Adams and 18 Eves"
- Archived data: Low Resolution RFLP Screening
- How to cite MITOMAP 



Map of the Human Mitochondrial DNA

About Mitomap

Human Mitochondrial DNA Revised Cambridge Reference Sequence
Last updated 03/26/2007

The rCRS sequence below is a corrected version of the original Cambridge Reference Sequence
This sequence is in GenBank as REFSEQ AC_000021.2 gi:115315570 and HUMMTCG J01415.2 gi:113200490.

This rCRS sequence replaces the 1997 HUMMTCG J01415.1 gi:1944628** and corrects the original 1981 CRS, J01415 gi:337188**.

Download this rCRS and other complete mtDNA genomes.

The rCRS sequence below represents the universally accepted rCRS of Anderson et al 1981 as revised by Andrews et al 1999. It differs from the original CRS and other complete hmtdna GenBank sequences in that it has eighteen annotated nucleotides. See the summary table of the reanalysis by Andrews et al.

- **Seven nucleotides are considered to be rare polymorphisms** and were determined to be correct as originally sequenced (J01415 gi:337188).
  Nucleotides **263A, 311C-315C, 750A, 1438A, 4769A, 8860A, and 15326A** are considered to be rare polymorphisms and are maintained as part of the true reference sequence. The seven rare polymorphisms are shown below in bolded green capitals.
- **Eleven nucleotide errors in the original CRS have been corrected** by re-sequencing the original placental material.
  Nucleotides **3107del***, **3423T**, **4985A**, **9559C**, **11335C**, **13702C**, **14199T**, **14272C**, **14365C**, **14368C**, **14766C** are corrections of the original Cambridge sequence. The errors in the original Cambridge sequence have been attributed to sequencing errors (8 instances) and to the inclusion of bovine (2 instances) or HeLa (1 instance) DNA. See summary table. Corrected sequencing errors are shown below in bold red underlined capitals.

  ***3107del** is maintained in this revised sequence with the gap represented by an 'N'. **THIS ALLOWS HISTORICAL NUCLEOTIDE NUMBERING TO BE MAINTAINED**.

The original CRS (HUMMTCG, J01415 gi:337188) contains the 7 confirmed rare polymorphisms, but not the subsequent 11 error corrections. The revision of 1997, HUMMTCG, J01415.1 gi:1944628 has 5 differences from the universally accepted rCRS (below) of Anderson et al 1981 + Andrews et al 1999 in that it does not have **750A, 3107del, 4985A, 11335C and **14766C** (1 of the rare polymorphisms and 4 of the error corrections). A PDF of the original Anderson et al 1981 Nature paper may be downloaded here.

The L-strand is shown. View double-stranded version. For strand composition asymmetry and an explanation of L-strand/H-strand terminology, click here.

```
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16561 atcacgatg
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For the strand asymmetry and an explanation of L-strand/H-strand terminology, click here.

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